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INSTITUTE FOR CLINICAL AND EXPERIMENTAL MEDICINE
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Opponent's opinion of the dissertation entitled "Identification and characterization of inherited kidney disease", author Kendrah Kidd, MSc., Department of Pediatrics and Inherited Metabolic Diseases, 1st Medical Faculty of Charles University

The present dissertation is designed in five separate parts. The introduction comprises 56 pages and is divided into 8 chapters, the review of the literature used 14 pages, followed by a commentary and a summary of her results on 6 pages. Finally, there are 5 publications for which the PhD student is the first author once and co-author four times. These papers have been published in reputable journals with impact factor proving their high level. This is also declared by other documented papers (27 in total) in the form of citations for which the PhD student is a co-author (impact factor 0.7-18.9).

In the initial eight chapters the issue of hereditary kidney diseases is clearly and comprehensibly described, focusing on autosomal dominant tubulointerstitial kidney disease (ADTKD)- clinical manifestations, history of development of knowledge of its genetic and pathophysiological background and diagnosis, including pictorial appendices. Already in this section, the unquestionable contribution of the scientific group, of which the PhD student is a member, to the detection of new genes (UMOD, REN, MUC1) underlying ADTKD and the elucidation of the pathophysiological mechanisms leading to the manifestation of this disease is documented. The essential role of the PhD student was the collection of data of the examined individuals, or patients, and their family members, the establishment and maintenance of a registry, including a website, at the international level (Charles University-Wake Forest Rare Inherited Kidney Disease - RIKD - Team Research Framework), and the subsequent analysis and interpretation of the data obtained. In addition, the PhD student has been instrumental in the methodology of acquiring, processing and transporting samples, establishing protocols for sample storage (biobank) and communicating with patients, their families and treating physicians.

In a separate section, the PhD student summarizes the outcomes of her work in a clear and specific way, which are in line with the defined objectives.

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Based on the molecular genetic methods used, and knowledge of laboratory and clinical markers, it was possible to establish the diagnosis of chronic kidney disease (ADTKD) and its phenotype, identify risk factors for progression, determine the prognosis and make a diagnosis in other family members, which is crucial for further specialized care and opens the possibility of specific-targeted therapy in the future.

The present work is unique and highly actual, as its results contribute substantially to understanding the nature of ADTKD as one of the diseases leading to irreversible renal failure, which has important clinical implications.

This dissertation, by its scope, content and results, fully demonstrates the author's aptitude for further independent creative scientific work and I recommend the award of the degree of "Ph.D." D. degree in Molecular and Cell Biology, Genetics and Virology.

I have two questions for the author:

1. Based on your current knowledge of the pathophysiological mechanisms, where would you see a therapeutic option to influence the progression of ADTKD to irreversible renal failure?
2. In which individuals would you recommend genetic testing targeting ADTKD?

Prague, 12.6.2024

MUDr. Silvie Rajnochová Bloudíčková, Ph.D., DBA

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